

dbGaP Study Release Notes



Release Notes for NHLBI TOPMed - NHGRI CCDG INSPIRE AF, phs001545.v2.p1

"NHLBI TOPMed - NHGRI CCDG: Intermountain INSPIRE Registry"

For any questions or comments, please contact: dbgap-help@ncbi.nlm.nih.gov.

April 22, 2020 Version 1 Data set release date
July 23, 2021 Version 2 Data set release date

2021-07-23

Version 2 Data set release for NHLBI TOPMed - NHGRI CCDG INSPIRE AF now available

This release includes the addition of Freeze 9 whole genome sequences (WGS) and corresponding VCFs. Please refer to the latest study configuration report for a detailed description of each download component.

Consent group 1 (c1): Disease-Specific (Cardiovascular, metabolic disease and associated risk factors, MDS) (DS-MULTIPLE_DISEASES-MDS)

Data Type	subjects	samples
Phenotype	476	475
Seq_DNA_SNP_CNV(VCFs)	449	449
WGS	449	449

For a description of SAMPLE_USE terms, please see:

<https://www.ncbi.nlm.nih.gov/projects/gap/submission/GetSampleUseTypes.cgi>

Study and Phenotype Updates

1. New Study Accession

NHLBI TOPMed - NHGRI CCDG INSPIRE AF version 1 phs001545.v1.p1 has been updated to version 2. The dbGaP accession for the current data is **phs001545.v2.p1**. The participant number (p#) has not changed in version 2. No new subjects have been added to this study.

2. There are no updates to the phenotype datasets.

Molecular Data

- Two genomics accessions, phg001384.v1 and phg001608.v1 with freeze8 and freeze9 genotypes, respectively, are associated with the study.
- See download components 'sample-info' for manifest of genotyped samples and files.
- Genotype calls are available in matrix format ('genotype-calls-vcf') as VCF file(s) with samples of same data sharing consent.
- Quality control data are in 'genotype-qc' tars.

phg	freeze	sample_cnt	subject_cnt
phg001384.v1	8	449	449
phg001608.v1	9	449	449

Authorized Access (Individual Level Data)

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Individual level data are available for download through the dbGaP Authorized Access System upon approval of the Data Access Request (DAR):

- <https://dbgap.ncbi.nlm.nih.gov/aa/wga.cgi?login=&page=login>

Public FTP site (Summary Level Data Only)

All data tables, data dictionaries, and documents will be housed under one directory for ease of downloading. The data_dict filenames have an added study version number (phs#.v#) and deleted participant set number (p#) from the table accession (pht#.v#). The var_report filenames have an added study version number (phs#.v#). In the var_report files, variables contain version numbers (phv#.v#) and summaries were created for each consent group (c#). These FTP files are available at:

- <https://ftp.ncbi.nlm.nih.gov/dbgap/studies/phs001545/phs001545.v2.p1>

2020-04-22

Version 1 Data set release for NHLBI TOPMed - NHGRI CCDG INSPIRE AF now available

This release includes the addition of Freeze 8 whole genome sequences (WGS) brokered through the Sequence Read Archive (SRA), and VCFs derived from WGS. Please refer to the latest study configuration report for a detailed description of each download component.

Consent group 1 (c1): Disease-Specific (Cardiovascular, metabolic disease and associated risk factors, MDS) (DS-MULTIPLE_DISEASES-MDS)

Data Type	subjects	samples
Phenotype	476	475
Seq_DNA_SNP_CNV (VCFs)	449	449
WGS*	449	449

*These data are brokered through the Sequence Read Archive (SRA). Please see Authorized Access instructions below.

For a description of non-SRA SAMPLE_USE terms, please see:

<https://www.ncbi.nlm.nih.gov/projects/gap/submission/GetSampleUseTypes.cgi>

Molecular Data

1. See download components phg001384.v1.TOPMed_CCDG_INSPIRE_AF.sample-info.MULTI.tar for manifest of genotyped samples and files.
2. Genotype calls are available in matrix format ('genotype-calls-vcf') as VCF file(s) with samples of same data sharing consent.
3. Quality control data are in phg001384.v1.TOPMed_CCDG_INSPIRE_AF.genotype-qc.MULTI.tar.

Authorized Access (Individual Level Data and SRA Data)

Individual level data and Sequence Read Archive (SRA) data are available for download through the dbGaP Authorized Access System upon approval of the Data Access Request (DAR):

- <https://dbgap.ncbi.nlm.nih.gov/aa/wga.cgi?login=&page=login>

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Public FTP site (Summary Level Data Only)

All data tables, data dictionaries, and documents will be housed under one directory for ease of downloading. The data_dict filenames have an added study version number (phs#.v#) and deleted participant set number (p#) from the table accession (pht#.v#). The var_report filenames have an added study version number (phs#.v#). In the var_report files, variables contain version numbers (phv#.v#) and summaries were created for each consent group (c#). These FTP files are available at:

- <https://ftp.ncbi.nlm.nih.gov/dbgap/studies/phs001545/phs001545.v1.p1>